Abstract

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Name of Bachelor´s Thesis: Hereditary causes of thromboembolic diseases and their laboratory diagnosis.

The term thrombophilia is used to designation hereditary and acquired prothrombotic states in arteries, veins and microcirculation. A thrombus formation may lead to partial or complete closure of the blood vessels - deep vein thrombosis. Then, a gore can be released and subsequently travels through blood vessels and a heart, after passing close one of pulmonary arteries, resulting in a pulmonary embolism. This state is known as venous thromboembolic disease. Then, the gore can be released and subsequently travels through blood vessels and after passing through a heart a gore may result either in a pulmonary arteries, resulting in a pulmonary embolism.

The aim of the thesis is to describe methods used for a diagnostics of thrombophilia and genetically dependent causes of thromboembolic disease. Early and accurate diagnostics of thrombophilia plays an important role in a prevention of thromboembolic events and complications associated with them.

Currently, a large variety of molecular genetic markers is used with clearly demonstrated thrombophilia risk - mutation of factor V 1691G>A and mutation of prothrombin gene 20210G>A. A diagnostics of other polymorphisms associated with thromboembolism is indicated only in selected cases, but even a diagnostics of the mutation of factor V 1691G> A and the gene for prothrombin 20210G> A are indicated only selectively. A detection of polymorphisms metylentetrahydrofolate reductase, plasminogen activator inhibitor, all coagulation inhibitors, lipoprotein (a) and a number of other polymorphisms is indicated only in special cases.

From the methodological point of view, all methods of direct DNA diagnostics are based on the principle of the polymerase chain reaction. At present, a diagnostics of mutations is mainly used fragment analysis, real-time and multiplex PCR.